Human germ cell formation in xenotransplants of induced pluripotent stem cells carrying X chromosome aneuploidies.

Journal: Sci Rep

Publication Year: 2014

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PubMed link: 25242416

Funding Grants: Stanford CIRM Training Program

Public Summary:

Scientific Abstract:
Turner syndrome is caused by complete or partial loss of the second sex chromosome and is characterized by spontaneous fetal loss in >90% of conceptions. Survivors possess an array of somatic and germline clinical characteristics. Induced pluripotent stem cells (iPSCs) offer an opportunity for insight into genetic requirements of the X chromosome linked to Turner syndrome. We derived iPSCs from Turner syndrome and control individuals and examined germ cell development as a function of X chromosome composition. We demonstrate that two X chromosomes are not necessary for reprogramming or maintenance of pluripotency and that there are minimal differences in gene expression, at the single cell level, linked to X chromosome aneuploidies. Formation of germ cells, as assessed in vivo through a murine xenotransplantation model, indicated that undifferentiated iPSCs, independent of X chromosome composition, are capable of forming germ-cell-like cells (GCLCs) in vivo. In combination with clinical data regarding infertility in women with X chromosome aneuploidies, results suggest that two intact X chromosomes are not required for human germ cell formation, qualitatively or quantitatively, but rather are likely to be required for maintenance of human germ cells to adulthood.